Precision Medicine, past, present and future

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At the beginning of this century public and private efforts generated a draft sequence of the human genome. The complete human genome sequence was published a few years later. This DNA sequence enabled the scientific community to better understand the genetic make up of humans and enabled the rapid cloning of many human disease genes. During the past 20 years, sequencing the whole genome, whole exome or the coding sequences of subsets of genes has become less expensive. In some parts of the world, clinical whole genome sequencing is now available at less than a $1,000 US. As the cost of DNA sequencing was going down, it was possible to sequence genomes from individuals across the world. An immediate impact of that sequencing was the identification of genomic variants and the beginning of an understanding of the role of such variants on health and disease. It is now well established that, in addition to rare Mendelian disorders, many common disorders also have a genetic basis.

Technologies for sequencing DNA, RNA and measuring proteins and metabolites have also evolved. As consequence, rapid and accurate methods of molecular diagnostics became available. It is now possible to assess risk of individuals and families to have children with genetic defects. Using non-invasive methods, it is possible to determine if an unborn fetus is carrying chromosomal abnormalities. Prior to these rapid sequencing technologies, many childhood disorders were diagnosed by the clinical symptoms and in some cases accurate diagnosis required years of waiting. Sequencing now permits rapid and accurate diagnosis of many of these disorders.

In addition to germ-line testing, Precision Medicine found a niche in diagnosing and treating cancer patients. Prior methods of classifying tumors were based on the tissue of origin and histology of tumor tissue. Large scale efforts to sequence tumor genomes revealed that each tumor type is heterogenous and each subtype can be defined on the basis of mutations or alternations in specific genes. Pharmaceutical and biotechnology companies developed drugs to target tumors with specific genetic/genomic alterations. Administration of drugs based on the genetic profile of individual tumors is changing the response rates to targeted therapies and, in some cases, the long-term survival of patients.

Use of genetic and genomic information to diagnose, assess the prognosis and to treat patients has now been defined as Precision medicine. Precision medicine is becoming an integral part of many medical specialties and implementation of the principles of precision medicine is going to improve the health of human populations around the world.